myotonia congenita

Myotonia congenita is a disorder that affects muscles used for movement (skeletal muscles). Beginning in childhood, people with this condition experience bouts of sustained muscle tensing (myotonia) that prevent muscles from relaxing normally. Although myotonia can affect any skeletal muscles, including muscles of the face and tongue, it occurs most often in the legs. Myotonia causes muscle stiffness that can interfere with movement. In some people the stiffness is very mild, while in other cases it may be severe enough to interfere with walking, running, and other activities of daily life. These muscle problems are particularly noticeable during movement following a period of rest. Many affected individuals find that repeated movements can temporarily alleviate their muscle stiffness, a phenomenon known as the warm-up effect.

The two major types of myotonia congenita are known as Thomsen disease and Becker disease. These conditions are distinguished by the severity of their symptoms and their patterns of inheritance. Becker disease usually appears later in childhood than Thomsen disease and causes more severe muscle stiffness, particularly in males. People with Becker disease often experience temporary attacks of muscle weakness, particularly in the arms and hands, brought on by movement after periods of rest. They may also develop mild, permanent muscle weakness over time. This muscle weakness is not seen in people with Thomsen disease.

Frequency

Myotonia congenita is estimated to affect 1 in 100,000 people worldwide. This condition is more common in northern Scandinavia, where it occurs in approximately 1 in 10,000 people.

Genetic Changes

Mutations in the *CLCN1* gene cause myotonia congenita.

The *CLCN1* gene provides instructions for making a protein that is critical for the normal function of skeletal muscle cells. For the body to move normally, skeletal muscles must tense (contract) and relax in a coordinated way. Muscle contraction and relaxation are controlled by the flow of charged atoms (ions) into and out of muscle cells. Specifically, the protein produced from the *CLCN1* gene forms a channel that controls the flow of negatively charged chlorine atoms (chloride ions) into these cells. The main function of this channel is to stabilize the cells' electrical charge, which prevents muscles from contracting abnormally.

Mutations in the *CLCN1* gene alter the usual structure or function of chloride channels. The altered channels cannot properly regulate ion flow, reducing the movement of

chloride ions into skeletal muscle cells. This disruption in chloride ion flow triggers prolonged muscle contractions, which are the hallmark of myotonia.

Inheritance Pattern

The two forms of myotonia congenita have different patterns of inheritance. Thomsen disease is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition.

Becker disease is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Most often, the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but do not show signs and symptoms of the condition.

Because several *CLCN1* mutations can cause either Becker disease or Thomsen disease, doctors usually rely on characteristic signs and symptoms to distinguish the two forms of myotonia congenita.

Other Names for This Condition

- Congenital myotonia

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Congenital myotonia, autosomal dominant form
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2936781/
- Genetic Testing Registry: Congenital myotonia, autosomal recessive form
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0751360/
- Genetic Testing Registry: Myotonia congenita
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0027127/

Other Diagnosis and Management Resources

- GeneReview: Myotonia Congenita
  https://www.ncbi.nlm.nih.gov/books/NBK1355
- MedlinePlus Encyclopedia: Myotonia congenita
  https://medlineplus.gov/ency/article/001424.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html

- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Myotonia congenita
  https://medlineplus.gov/ency/article/001424.htm
- Health Topic: Muscle Disorders
  https://medlineplus.gov/muscledisorders.html

Genetic and Rare Diseases Information Center

- Myotonia congenita autosomal dominant
  https://rarediseases.info.nih.gov/diseases/6176/myotonia-congenita-autosomal-dominant
- Myotonia congenita autosomal recessive
  https://rarediseases.info.nih.gov/diseases/844/myotonia-congenita-autosomal-recessive

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke
  https://www.ninds.nih.gov/Disorders/All-Disorders/Myotonia-Information-Page

Educational Resources

- Disease InfoSearch: Myotonia congenita autosomal dominant
  http://www.diseaseinfosearch.org/Myotonia+congenita+autosomal+dominant/5073
- Disease InfoSearch: Myotonia congenita autosomal recessive
  http://www.diseaseinfosearch.org/Myotonia+congenita+autosomal+recessive/5074
- MalaCards: myotonia congenita
  http://www.malacards.org/card/myotonia_congenita
- Merck Manual Consumer Version: Congenital Myopathies
  http://www.merckmanuals.com/home/children-s-health-issues/muscular-dystrophies-and-related-disorders/congenital-myopathies
- Orphanet: Thomsen and Becker disease
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=614

Patient Support and Advocacy Resources

- Muscular Dystrophy Association
  https://www.mda.org/disease/inherited-and-endocrine-myopathies
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/myotonia-congenita/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/muscular.html

GeneReviews

- Myotonia Congenita
  https://www.ncbi.nlm.nih.gov/books/NBK1355

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22myotonia+congenita%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Myotonia+Congenita%5BMAJR%5D%29+AND+%28%28myotonia+congenita%5BTIAB%5D%29+OR+%28Becker's+myotonia%5BTIAB%5D%29+OR+%28Thomsen's+disease%5BTIAB%5D%29+OR+%28Thomsen+disease%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- MYOTONIA CONGENITA, AUTOSOMAL DOMINANT
  http://omim.org/entry/160800
- MYOTONIA CONGENITA, AUTOSOMAL RECESSIVE
  http://omim.org/entry/255700

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/myotonia-congenita

Reviewed: April 2007
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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Department of Health & Human Services